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Clinical, morphological and genetic features of a cohort of late onset GSD II patients: typical and atypical presentations

E Barca*, O Musumeci, C Rodolico, A Ciranni, G Vita, A Toscano

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Introduction

GSDII (Pompe disease) is a rare, autosomal recessive disease due to alpha-glucosidase (GAA) deficiency that presents with infantile and late-onset forms. Herein, we describe a cohort of 29 late-onset patients (15 males and 14 females, aged 9 to 76), and discuss some unusual clinical features seen in 4 of them.

Methods

Clinical evaluations were performed in all patients using the Walton and Gardner-Medwin, MRC and GSGC scales, and the 6MWT. Respiratory function was assessed using FVC% in upright and supine positions. All patients underwent morphological and biochemical examinations, muscle MRI, neurophysiological studies, and molecular genetic analysis.

Results

Out of 29 patients, 15 received enzyme replacement therapy (ERT). 57% initially had limb-girdle involvement, 29% an isolated hyperCKemia, and 14% respiratory insufficiency. Neurophysiological studies revealed a myopathic pattern in 48% of patients, neurogenic or mixed in 28%, whereas 24% of patients did not show any electrical abnormality. In the examined patients, muscle MRI showed early involvement of ileopsoas, gluteus and posterior thigh muscles. Morphological studies revealed vacuolar myopathy in 68% of patients, and biochemical analysis showed residual alpha-glucosidase activity ranging from 0.01% to 25%. Molecular genetic analysis confirmed IVS13T>G as the most common mutation, but we also identified three novel point mutations. Atypical clinical features were observed in 4 out of 29 patients: a 66-year-old female with disease onset involving distal upper limbs muscles; a 70-year-old male with mesial temporal sclerosis without epileptic or cognitive disorders; a 46-year-old male with severe neurosensory hearing loss (with very early hearing difficulties); and a 51-year-old female with a congenital absence of a thyroid lobe.

Conclusion

Our study confirms the clinical, morphological and molecular genetic heterogeneity of late-onset GSDII. Muscle MRI is a very useful tool for muscle damage evaluation, especially at the early stages of the disease. Based on our experience with some atypical cases, we suggest always performing a complete clinical and laboratory evaluation of patients in order to highlight unusual muscular (e.g. distal involvement) and/or CNS presentations. These cases reinforce the hypothesis that modifier genes and/or epigenetic factors may contribute to clinical presentations of Pompe disease.

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Department of Neurosciences, University of Messina, Messina, Italy